

# Angels at Bethesda



Charitable medical air transportation serving patients of the National Institutes of Health

## Angel Flight Helps Family with Eye Disease

**M**ario C., of Hollywood, Fla., including his cousins, Ricardo and Kung-Fu (cautiously), but other suffers from a genetic eye disorder known as X-linked Juvenile Retinoschisis (RS) that causes poor eyesight and degeneration of the retina. William of New York; and a cousin, activities such as baseball are Nelson, from Chicago, who is legally prohibited because of the danger of blind. Two young nephews, Daniel, 5, retinal detachment.



Doctors can't fix 'boo-boo' in brothers' eyes

Retinoschisis (RS) that causes poor eyesight and degeneration of the retina.

The disease affects only males and is carried in the gene of the mother, who remains unaffected.

A native of Colombia, Mario took an Angel Flight to the National Eye Institute in January for an evaluation, calling the trip "a great experience. The pilots were wonderful."

Other men in Mario's family suffer from RS as well, and have taken charitable flights to NIH for testing,

and Andrew, 2, traveled by car with their parents.

Symptoms of RS present differently. One of Mario's cousins can't see directly in front of himself. Another views a person as a puzzle. Still another, whose vision is only peripheral, tilts his head sideways. There is no cure, but NIH is working to find one and has had positive results in animal testing.

Daniel and Andrew's mom, Maria C., says her children will return to NIH when they call to say "we have a cure."

Once treatment for humans becomes available, the boys will participate in a clinical trial. "They'll be the first ones to use the medicine," Maria said.

But for now, life remains challenging. "Daniel told me the doctors 'can't fix the boo-boo in our eyes.'"

Daniel plays soccer and takes

The boys' uncle, Mario, says that as he gets older, his vision will further deteriorate. "I'm not getting worse. I'm not getting better. I live a normal life. I drive. It's hard for me to read



Ricardo and William with half-brother Dario (L to R) share a good time on New Year's Eve. Since his mother is not a carrier of RS, Dario's vision is normal.

for long—I use a magnifying glass."

Mario is the manager of a McDonalds in Hollywood. He and his wife Maria Elena have two daughters, Natalie, 16, and Caroline, 12.

"The little one has been carrying this idea of becoming an eye doctor for years now."

## Serving Rare Disease Community Is Priority



**Ed Boyer**

I am extremely pleased that Angel Flight can serve patients and families with rare diseases. Not only do I serve as the chairman of Angel Flight America, I also am on the board of directors of the National Organization of Rare Disorders (NORD).

I know the importance of serving the rare disease community. Everybody needs to understand that charitable or charitably-assisted patient transportation is available to all within the limits of available resources.

*Ed Boyer is the CEO and president of Angel Flight of Virginia, and a retired DHHS-OS employee.*

### NIH Missions January 2004 to March 2006

<b>Missions</b>	<b>161</b>
<b>Passengers</b>	<b>282</b>
<b>Patients</b>	<b>90</b>

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## Cystinosis No Hindrance to 3-Year-Old

Looking at vivacious, curly-haired Channing, you would never know the three-year-old suffers from a rare metabolic disorder called cystinosis that in most patients causes renal failure, poor eyesight, and retarded growth.

She takes tap, guitar, ballet, eats constantly, and loves spicy-hot food.

Channing's exceptional rates of growth and development caught the attention of Dr. William Gahl, clinical director of the National Human Genome Research Institute and an expert in cystinosis at NIH. He requested that Channing come to NIH for evaluation. Marita Eddy, transportation coordinator for Angel Flight in Bethesda, arranged for Channing and Lanna, her mom, to fly from Gainesville, Fla., on Nov. 14, for five days of testing at NHGRI. Eddy said the little girl is "quite the performer."

Diagnosed at 10 months with the disease, which is caused by the accumulation of the amino acid cystine in various organs of the body, Channing takes Cystagon every six hours.

Cystagon effectively dissolves the crystals that can form in the kidneys, eyes, pancreas, and brain. Lanna says diligence is critical in administering the medicine. Without treatment, most children experience end-stage renal failure and must receive a transplant. Many require a feeding tube (the disease used to be called "renal dwarfism" because the affected children failed to grow and thrive). Eighty percent of these children will have it throughout their entire lives, Lanna says. One doctor wanted to put Channing on a feeding tube, but Lanna resisted. "Let's try the meds first," she said.

As a result of taking Cystagon consistently as prescribed, Channing has exceeded all expectations for a child with cystinosis. "Dr. Gahl couldn't believe she was 90 percent on the growth chart. Most kids aren't even in the five percent category," Lanna says. Dr. Gahl will be following Channing's progress throughout her whole life.

"Cystinosis is not easy, especially at the beginning," notes the former dancer for Disneyworld. "I haven't slept more than six hours at a time since Channing was ten months old. Some parents grow lax, citing their careers and marriages as priorities, but their children pay the price with worsening symptoms. If I could win the lottery, I would buy a private jet and fly around the world educating and talking to parents. A feeding tube is not the best alternative."

Flying with Angel Flight was "100 percent a great experience," Lanna says. "Channing was really good. She'd never been on a plane before. The pilots were great."



**Channing is 'quite the performer'**